Galactosemia
General Overview

Q. What is galactosemia?

A. Galactosemia is a treatable disorder. It affects the way the body processes the sugar galactose, a component of milk and dairy products. Children with galactosemia cannot process galactose. As a result, galactose and other by-products build up in the bloodstream and cause physical and developmental damage.

Q. How does the body normally process galactose?

A. The body normally converts galactose into glucose, which is used to fuel the body. This conversion is made possible by several enzymes. One of these, named galactose-1-phosphate uridyltransferase (GALT), is associated with galactosemia.

Q. What happens to galactose in a child with galactosemia?

A. In a child with galactosemia, galactose cannot be converted to glucose because the GALT enzyme does not work properly. This results in large amounts of galactose and other by-products that are toxic to the body.

Q. What are the effects of having galactosemia if it is not treated?

A. Untreated galactosemia can cause rapid, unexpected death due to an infection that invades the blood. Infants with untreated galactosemia may also develop brain damage, liver disease, and cataracts. Each child with galactosemia is different so the outcome will not be the same for all children.

Q. What is the treatment for galactosemia?

A. The first step in treatment is to remove the baby from breast milk and commercial formula. Soy-based formulas that contain no galactose should be substituted into the baby's diet. Individuals with galactosemia must remain on a restricted diet and maintain low blood galactose levels throughout life.

Q. Is there only one form of galactosemia?

A. No, there are several forms of galactosemia. The most severe form of galactosemia is caused by absence or inactivity of GALT.

Q. Why would a child have galactosemia?

A. Galactosemia is an inherited disorder. It results when a baby receives a non-working GALT gene from each parent. For more information about this, contact your health care provider or a genetic counselor.

Q. How common is galactosemia?

A. About one in every 50,000 babies in the United States is born with galactosemia.

For more information about galactosemia, please see the Disorders section of our website: www.doh.wa.gov/nbs

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